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The genetics of familial hypercholesterolemia and its detection

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Familial Hypercholesterolaemia is an autosomal, dominant genetic disorder that leads to elevated blood cholesterol and a dramatically increased risk of atherosclerosis. It is perceived as a rare condition. However it affects 1 in 250 of the population globally, making it an important public health concern. In communities with founder effects, higher disease prevalence's are observed. We discuss the genetic basis of familial hypercholesterolaemia, examining the distribution of variants known to be associated with the condition across the exons of the genes LDLR, ApoB, PCSK9 and LDLRAP1. We also discuss screening programmes for familial hypercholesterolaemia and their cost-effectiveness. Diagnosis typically occurs using one of the Dutch Lipid Clinic Network (DCLN), Simon Broome Register (SBR) or Make Early Diagnosis to Prevent Early Death (MEDPED) criteria, each of which requires a different set of patient data. New cases can be identified by screening the family members of an index case that has been identified as a result of referral to a lipid clinic in a process called cascade screening. Alternatively, universal screening may be used whereby a population is systematically screened. It is currently significantly more cost effective to identify familial hypercholesterolaemia cases through cascade screening than universal screening. However, the cost of sequencing patient DNA has fallen dramatically in recent years and if the rate of progress continues, this may change.

Biography

Victor Kerchev and his co-authors has its own experience in evaluating and passion to improve the health and well-being of cardiological patients. The authors open and contextual assessment model based on reacting Constructivists creates new ways to improve the medical care of patients with the familial form of hypercholesterolemia. They created the model after many years of experience in research, evaluation and training in hospitals and schools. This approach responds to all parties concerned, and has a different focusing method.

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